

CLAIMS

I claim:

1. A method for the determination of data for the preparation of presymptomatic or prenatal diagnosis of phakomatosis comprising the steps:

a. amplifying a polymorphous DNA microsatellite marker from a tumor of an afflicted individual who is suffering from the phakomatosis,

b. amplifying the polymorphous DNA microsatellite marker from the blood of the afflicted individual, and

c. comparing the length of the amplified polymorphous DNA microsatellite markers from steps (a) and (b).

2. The method according to claim 1, further comprising the steps of amplifying the polymorphous DNA microsatellite marker from the offspring of afflicted individual and comparing the length of the amplified marker with the length of the amplified polymorphous DNA microsatellite markers from steps (a) and (b).

3. The method according to claim 1 or 2, further comprising the steps of amplifying two or more different polymorphous DNA microsatellite markers from the tumor and the blood.

4. A method for the determination of data for the preparation of presymptomatic or prenatal diagnosis of phakomatosis comprising the following steps:

a) making available a tumor material of an afflicted individual who is suffering from the tumor suppressor gene disease,

- b) making available the blood of the afflicted individual,
- c) isolating the tumor DNA from the tumor material,
- d) isolating the blood DNA from the blood,
- e) amplifying at least two polymorphous DNA microsatellite markers from the tumor material,
- f) amplifying the polymorphous DNA microsatellite marker from the blood,
- g) separating by length the polymorphous DNA microsatellite markers from the tumor material,
- h) separating by length the polymorphous DNA microsatellite markers from the blood,
- i) observing the length of the polymorphous DNA microsatellite markers from the tumor material,
- j) observing the length of the polymorphous DNA microsatellite markers from the blood, and
- k) determining the allele which is lost in the tumor.

5. The method according to claim 1, further comprising the steps of amplifying the same polymorphous DNA microsatellite marker from the blood of the offspring of the afflicted individual and comparing the length of the amplified marker with the length of the amplified polymorphous DNA microsatellite markers from steps (e) and (f).

6. The method according to any one of claims 1-4, wherein the phakomatosis is a tumor suppressor gene disease.

7. The method according to claim 6, wherein the phakomatosis is a neurofibromatosis.

8. The method according to any one of claims 1-7, where the two polymorphous markers, of which there are at least two, preferably have a length of up to approximately 300 bp.

9. The method according to any one of claims 1-8, wherein at least three, or preferably four, different markers are used.

10. The method according to any one of claims 1-9, wherein the marker is a neurofibromatosis gene flanking or intragenic marker.

11. The method according to claim 10, wherein at least one of the markers is located in intron 27 of the neurofibromatosis type 1 gene.

12. The method according to claim 10, wherein at least one of the markers is located in intron 38 of the neurofibromatosis type 1 gene.

13. The method according to claim 10, wherein, the marker is selected from the group consisting of CRYB2, D22S275, NF2CA3, D22S268 and D22S430.

14. The method according to any one of claims 1-7, wherein the afflicted individual is a parent of the offspring.

15. The method according to any one of claims 1-7, further comprising repeating steps d), f), h) and j).

16. The method according to any one of claims 1-7, further comprising repeating steps c), e), g) and i).

17. The method according to any one of claims 1-7, further comprising repeating steps c), e), g) and i) with at least one additional tumor of the afflicted individual.

18. The method according to any one of claims 1-7, further comprising the steps of amplifying the polymorphous DNA microsatellite marker from the blood of an unaffected relative of the offspring and observing the amplified marker DNA.